

# Final Programme – SASHG 2015

6 Aug 2015

Sunday, 16 August 2015			
10h00	11h30	<b>Genetic Counsellors Workshop</b> Facilitator: Dawn Allain	<b>Ethical dilemmas in genetics: A framework to address dilemmas raised by new technologies</b>
12h00	13h00	Genetic Counsellors-South Africa - AGM	
<b>12h00</b>	<b>18h00</b>	<b>REGISTRATION</b>	
<b>14h00</b>	<b>14h40</b>	<b>OPENING SESSION</b>	<b>CHAIR: Lizette van Rensburg</b>
14h00	14h10	Chair SASHG – Amanda Krause	Opening and welcome
14h10	14h30	TBA	Opening Address
14h30	14h40	Chair Organizing Committee – Lizette van Rensburg	
14h40	15h00	<i>Comfort break</i>	
<b>15h00</b>	<b>17h00</b>	<b>Southern African Human Genome Programme</b>	<b>CHAIRS: Michèle Ramsay &amp; Raj Ramesar</b>
15h00	15h20	Rizwana Amod	Overview of current developments in Science & Technology of relevance to health research
15h20	15h40	Michèle Ramsay / Michael Pepper	The design of the SAHGP - its history and logistical issues
15h40	16h00	Shaun Aron	Summary of findings and interesting insights from data analyses
16h00	16h20	Ananyo Choudhury	Understanding population structure and a way forward
16h20	16h40	Himla Soodyall	Ethical, Legal Social Issues: lessons learnt in SA Population Studies - the way forward
16h40	17h00	Claudette Medefindt	A consumers point of view: Genomics for people living with hereditary conditions
<b>17h00</b>	<b>18h00</b>	<b>POSTER VIEWING - SESSION 1</b>	
<b>18h00</b>	<b>20h00</b>	<b>WELCOME COCKTAIL PARTY</b>	
Monday, 17 August 2015			
<b>7h30</b>	<b>16h00</b>	<b>REGISTRATION</b>	
<b>8h30</b>	<b>10h10</b>	<b>MENDELIAN DISORDERS / MOLECULAR PATHOLOGY</b>	<b>CHAIR: Engela Honey</b>
8h30	9h10	<b>Keynote: Bart Loeys</b>	<b>An overview of the genetics of thoracic aorta dissections</b>
9h10	9h25	Nadia Carstens	Novel mutation in the CHST6 gene causes macular corneal dystrophy in a black South African family
9h25	9h40	Elana Vorster	Determining the molecular basis of spinal muscular atrophy (SMA) in the black South African population
9h40	9h55	Thandiswa Ngcungcu	Non-coding variants segregate with disease in South African families with Keratolytic Winter Erythema (KWE)
9h55	10h10	Celia van der Merwe	Curcumin shows protective effects in a PINK1 siRNA-mediated model of Parkinson's disease
<b>10h10</b>	<b>10h40</b>	<b>TEA BREAK</b>	

<b>10h40</b>	<b>12h10</b>	<b>GENETIC EPIDEMIOLOGY / POPULATION CHAIR: Himla Soodyal</b>	
10h40	10h55	Danielle Smith	Beyond our borders: evidence for a Spinocerebellar ataxia type 7 founder effect in southern Africa
10h55	11h10	Cheryl Stewart	Finger on the pulse – The utility of establishing a national CF registry
11h10	11h25	Jeanne van Rensburg	Cystic fibrosis and rarefaction: Unification through diversity
11h25	11h40	Melissa Nel	The African-specific -387 C>T TGFB1 Promoter Polymorphism is functional and associates with an 'African complication' in juveniles with Myasthenia Gravis
11h40	11h55	Mario Moller	Population admixture: friend or foe in TB susceptibility
11h55	12h10	Caitlin Uren	Investigating fine-scale population structure between the Nama and ≠Khomani San of South Africa
<b>12h10</b>	<b>13h10</b>	<b>POSTER VIEWING - SESSION 2</b>	
<b>13h00</b>	<b>14h00</b>	<b>LUNCH BREAK</b>	<i>Personalized genomics using a comprehensive suite of gene panels for rare inherited diseases : Dr Alain Rico, Genetic Sciences Division, Thermo Fisher Scientific</i>
<b>14h00</b>	<b>14h45</b>	<b>COMPLEX DISORDERS CHAIR: Louise Warnich</b>	
14h00	14h15	Hadassa Goldfein	Associations between mtDNA haplogroup variation and sensory neuropathy in South Africans using d4T
14h15	14h30	Nathaniel McGregor	The identification of novel genes in anxiety disorders: a gene x environment correlation and interaction study
14h30	14h45	Melvin AnyasiAmbele	Genome-wide analysis of gene expression during adipogenesis in human adipose-derived mesenchymal stromal cells (ASCs)
<b>14h45</b>	<b>15h20</b>	<b>TEA BREAK</b>	<i>Your new research companion for Pharmacogenomics: Dr. Peter Jacobs, Genetic Sciences Division, Thermo Fisher Scientific</i>
15h30	17h00	SASHG Biennial General Meeting	
<b>18h00</b>	<b>22h00</b>	<b>DINNER</b>	<b>Social at Klein-Kaap</b>
<b>Tuesday, 18 August 2015</b>			
<b>7h30</b>	<b>16h00</b>	<b>REGISTRATION</b>	
<b>8h30</b>	<b>10h10</b>	<b>Association Studies CHAIR: Zane Lombard</b>	
8h30	9h10	<b>Keynote: John Kemp</b>	<b>Genetic determinants of bone mineral density and osteoporosis</b>
9h10	9h25	Tinashe Chikowore	Evaluation of common variants associated with Type 2 Diabetes in the Asian and European ethnicities among the Black South African population of Tswana descent
9h25	9h40	Liesl Hendry	The genetics of blood pressure in black South African individuals from the Birth to Twenty cohort
9h40	9h55	Venessa Pillay	The MetaboChip as a tool for identifying genetic markers of obesity risk in a South African black population
9h55	10h10	Ellen Ovenden	Investigating the functional significance of genome-wide variants associated with treatment response in schizophrenia
<b>10h10</b>	<b>10h40</b>	<b>TEA BREAK</b>	

<b>10h40</b>	<b>12h20</b>	<b>NEXT GENERATION SEQUENCING / BIOINFORMATICS CHAIR: Soraya Bardien-Kruger</b>	
10h40	11h20	<b>Keynote: Chris Mathew</b>	<b>Next generation sequencing for detection of germline and somatic variants involved in gastrointestinal disease</b>
11h20	11h35	Brigitte Glanzmann	Next generation sequencing methods to identify pathogenic mutations implicated in human diseases: Holy Grail or tin cup?
11h35	11h50	Nicole van der Merwe	Functional DNA mismatch repair variant missed in exomes of BRCA-negative breast cancer patients using variant calling with the public human genome reference sequence (hg19)
11h50	12h05	Lisa Roberts	Whole exome sequencing to address the missing heritability of inherited retinal diseases in indigenous Africans
12h05	12h20	Shareefa Dalvie	Bipolar Disorder: Whole-genome sequencing in a large Afrikaner family
<b>12h20</b>	<b>13h20</b>	<b>POSTER VIEWING - SESSION 3</b>	
<b>13h15</b>	<b>14h15</b>	<b>LUNCH BREAK</b>	<i>CMA in reproductive health : Ms Lotta Olofsson, Clinical Sales Specialist Nordic countries, Affymetrix UK LTD</i>
<b>14h15</b>	<b>15h40</b>	<b>CLINICAL GENETIC/GENOMIC TESTING</b>	<b>CHAIR: Jacquie Greenberg</b>
14h15	14h55	<b>Keynote: Brynn Levy</b>	<b>Clinical utility of the new cytogenetic/cytogenomic tools in reproductive medicine</b>
14h55	15h10	Karen Fieggen	Outcome of targeted MLPA CNV testing at a paediatric genetic clinic in South Africa
15h10	15h25	Fahmida Essop	Unique SNPs found in the South African Black population can lead to misdiagnosis of common genetic tests
15h25	15h40	Craig Kinnear	Mutation Screening for Primary Immunodeficiency disorders in a tuberculosis endemic region: a South African perspective
<b>15h40</b>	<b>16h15</b>	<b>TEA BREAK</b>	<i>Sample to Insight: Analyzing and interpreting the biological meaning in human NGS data with ease - Dr Anne Arens – QIAGEN Advanced Genomics</i>
16h20	17h20	Clinical Genetics Focus Group - Meeting	
<b>18h00</b>		<b>Bus departs from Klein-Kaap for Royal Elephant / Bondev House - Gala Dinner</b>	
<b>19h00</b>	<b>23h30</b>	<b>GALA DINNER</b>	<i>Royal Elephant / Bondev House : Walking Tall performance, Prize giving</i>
<b>22h00</b>		<b>1<sup>st</sup> Bus to return to Klein-Kaap</b>	
<b>23h30</b>		<b>Last Busto return to Klein-Kaap</b>	
<b>Wednesday , 19 August 2015</b>			
<b>7h30</b>	<b>12h00</b>	<b>REGISTRATION</b>	
<b>8h30</b>	<b>9h55</b>	<b>GENETIC COUNSELLING / SUPPORT GROUPS CHAIR: Tina-Marie Wessels</b>	
8h30	9h10	<b>Keynote: Dawn Allain</b>	<b>Next generation sequencing panels and whole exome sequencing: Impact, challenges and lessons learned in clinical practice</b>
9h10	9h25	Chantelle Scott	Development of an informational video regarding prenatal diagnosis for Down syndrome at Tygerberg Hospital
9h25	9h40	Mardelle Schoeman	Genetic counselling for Non-Invasive Prenatal Testing: a report on 308 cases
9h40	9h55	<b>Jennifer Kromberg / Helen Malherbe</b>	The past, present and future of the Southern African Inherited Disorders Association

<b>10h00</b>	<b>10h30</b>	<i>TEA BREAK</i>	
<b>10h30</b>	<b>12h00</b>	<b>FUTURE PERSPECTIVESCHAIR: Lizette van Rensburg</b>	
10h30	11h00	<b>Plenary: ReinhardHiller</b>	<b>Precision medicine</b>
11h00	11h30	<b>Plenary: Bertram Henderson</b>	<b>The next generation: Social media and Medical Genetics</b>
11h30	11h45	<i>Jacque Greenberg</i>	New technologies, new challenges: guidelines for the ethical oversight of informed consent for the use of induced pluripotent stem cells for future research purposes
11h45	12h00	SASHG Chair	<i>Closing Remarks</i>
<b>12h00</b>	<b>13h00</b>	<i>Lunch break</i>	